Rapid Genome Sequencing (rGS)

Your child’s doctor has recommended a genetic test called rapid genome sequencing (rGS). This test looks for changes in DNA that could cause or contribute to a medical condition.

About Your Genetics

Our bodies are made up of billions of building blocks called cells. Each cell contains DNA. Small parts of DNA are called genes. Genes tell our body how to grow, develop, and function. There are about 20,000 genes in human DNA. The complete set of genes, or genetic material, in a cell or living thing is called a genome.

Changes in our DNA can have an impact on our health. Everyone is born with their own DNA changes. Some DNA changes cause or contribute to diseases, but most do not. Your child’s doctor thinks knowing more about your child's DNA changes will help them take better care of your child.

How the Test is Done

An rGS test looks for DNA changes in a blood or spit (saliva) sample. Blood is the best sample for rGS. If a saliva sample is used, testing may take longer to complete. Results of rGS are best understood when the lab has samples from your child and both biological parents. This helps the lab know which DNA changes came from the parents and which are new to your child. DNA changes that might explain your child’s medical condition are reported.

You can expect to get test results within 8 days.
Test Results

The results of this test can be:

- **Positive:** DNA change(s) that cause or contribute to your child’s medical condition is present. Positive results may tell the genetic team more about:
  - Your child’s diagnosis.
  - Some, or all, of the cause of your child’s symptoms.
  - What to expect and possible treatments.

- **Negative:** No DNA changes that cause disease are present. However, there could still be a genetic cause for your child’s medical condition that was not found by this test.

- **Uncertain:** Genetic changes are present that may or may not contribute to your child’s medical condition.

After the Test

Your child’s doctor and/or genetic counselor will talk to you about the test results. It is possible that more testing may be recommended. If you have questions, please ask.